

Appendix S1: Additional symptoms and features in patients with CMT reported in the literature.

Mutated gene	Severe sensory	Vocal cords	Tremor	UL predominant	Contractures	Scoliosis	Hand deformities	Skeletal	Deafness	Cognitive impairment	Bulbar
PMP22	-	-	+[1,2]	-	+[3]	+[1,2,4]	+[1,2]	cong. hip dysplasia, ulnar deviated hand, lordosis[5], chest deformity[6]	+[1,2]	+[7]	+[3,4]
MPZ	+[8]	+[9]	+[10]	-	+[11]	+[12,8,11]	+[8]	chest deformity[13]	+[12,14]	+	+[15,11]
GJB1	-	+[19]	+[20]	-	-	+[12]	-	-	+[12]	+[21]	+[21]
PRX	-	-	+[46]	-	-	+[46,47]	+[48]	kyphosis[49]	+[47]	-	-
EGR2	-	+[25,26]	+[27]	-	+[28]	+[29,30]	+[31,30]	hip dysplasia[32]	+[25]	-	+[32,26]
PLEKHG5	-	-	-	-	-	-	-	spine deformity[83]	-	-	-
MFN2	-	+[16]	+[16]	-	+[16]	+[16]	-	hyperlordosis, kyphosis[17]	+[16]	+[18]	+[168]
GDAP1	-	+[51]	-	-	+[51]	+[52]	+[51]	joint laxity[52], kyphosis[53], chest deformity[54]	-	-	+[52]
PDK3	-	-	+[74]	-	-	-	-	-	+[74]	-	-
SURF1	-	-	-	-	-	+[90]	-	-	+[90]	-	-
DHTKD1	-	-	-	-	-	-	-	-	-	-	-
INF2	-	-	+[65]	-	-	+[65]	+[65]	partial syndactyly[66], ulnar deviated hands, kyphosis	+[65]	+[66]	-
KIF1B	-	-	-	-	-	-	-	lordosis[75]	-	-	-
DYNC1H1	-	-	+[77]	-	-	-	-	lordosis[77]	-	+[77]	-
LMNA	-	-	-	-	-	+[78]	+[78]	hyperlordosis[79]	-	-	-
NEFL	+[22]	-	+[22]	-	+[22]	+[23]	+[24]	lordosis[23]	+[22]	+[22]	+[22]
TRIM2	-	-	-	-	-	-	-	-	-	-	-
GARS	-	-	-	+[40]	-	+[41]	-	-	-	-	-
KARS	-	-	-	-	-	-	-	-	-	-	-
AARS	-	-	-	-	-	-	-	-	+[76]	-	-
MARS	-	-	-	-	-	-	-	-	-	-	-
HARS	-	-	-	-	-	-	-	-	-	-	-
YARS	-	-	-	-	-	-	-	-	-	-	-
HINT1	-	-	-	-	-	-	-	-	-	-	-
LRSAMI	-	-	-	-	-	-	-	-	+[82]	-	-
SH3TC2	-	-	+[43]	-	-	+[43]	+[44]	-	+[43]	-	+[45]
RAB7	+[50]	-	-	-	-	+[50]	-	-	-	-	-
FIG4	-	-	+[60]	-	-	+[60]	+[61]	-	-	-	-
DNM2	-	-	-	-	-	-	-	-	-	+[64]	-
MTMR2	-	-	+[67]	-	-	+[68]	+[67]	chest deformity[67]	-	-	+[2]
TFG	-	-	+[69]	-	-	-	-	-	+[69]	-	+[69]
LITAF	-	-	+[70]	+	-	+[71]	-	-	+[71]	+	-
FGD4	+[80]	-	+[81]	-	-	+[81]	-	-	-	-	-
SBF2	-	-	-	-	-	+[86]	+[86]	-	+[87]	-	-
NDRG1	-	-	+[55]	-	-	+[55]	+[56]	-	+[55]	-	+[57]
HSPB8	-	-	-	-	-	+[42]	-	-	-	-	-
HSPB1	-	-	-	-	-	-	+[91]	-	-	-	-
HSJ1	-	-	-	-	-	-	-	-	-	-	-
MED25	-	-	-	-	-	-	-	-	-	-	-
JFRD1	-	-	+[92]	-	-	-	-	-	+[92]	-	+[92]
TRPV4	-	+[33]	+[34]	-	+[35]	+[34]	-	short stature[36], lordosis[37], cong. hip dislocation[38]	+[39]	-	+[37]
SLC12A6	-	-	+[88]	-	-	-	-	kyphosis[89], joint laxity[88]	-	+[89]	-
HK1	-	+[58]	+[58]	-	-	+[59]	+[58]	-	-	+[59]	-
AIFM1	-	-	-	-	-	-	-	-	+[62]	+[62]	-
PRPS1	-	-	-	-	-	-	-	-	+[63]	-	-
FBLN5	-	-	-	-	-	+[72]	+[73]	joint laxity, lordosis[72]	-	-	-
GNB4	-	-	-	-	-	-	-	-	-	-	-
SBFI	-	-	-	-	-	-	-	short stature, syndactyly and webbing[85]	-	+[85]	+[85]

Appendix S1 (continued): Additional symptoms and features in patients with CMT reported in the literature.

Mutated gene	Upper motor neuron	Fasciculations	Facial weakness	Early proximal weakness	Pain	Paresthesia	Early onset	Eye involvement	CTS	Respiratory
PMP22	-	+[5]	+[5,2]	-	+[5,3]	+[5]	+[5,3]	optic neuritis[93], pupill.[5,3]	+	diaphragmal weakness[5,2], respiratory failure , resp. distress at birth[3]
MPZ	-	+	+[94,95]	+[15,96]	+ [97,98]	+[97,98]	+[99,8, 11]	pupill.[12,98], ophthalmoparesis[94], optic atrophy[167]	+	resp. failure[97,100,95], chronic cough[101], resp. distress neonatal[15,111], resp. obstruction, stridor[9]
GJB1	+[21]	-	-	-	+	+[21]	+[20]	pupill.[102]	-	-
PRX	-	+ (to)[47]	-	+ [47]	-	+ [48,109]	+ [48]	glaucoma[110]	+ [46]	restr. resp. failure[110]
EGR2	-	+ [26]	+ [26]	+ [104]	-	-	+ [26]	ophthalmoparesis [25,26], strabismus[25,32,105], pupill.[26]	-	restrictive pulmonary disease, resp. failure (with death)[26]
PLEKHG5	-	-	-	-	-	-	-	-	-	-
MFN2	+[16]	-	+ [17]	+ [16]	+ [16]	+	+ [17]	bilat. optic atrophy[16], ophthalmoparesis [18], pupill.[102]	-	resp. failure[103]
GDAP1	-	-	+ [52]	-	+ [112]	-	+ [52]	optic atrophy[112]	-	diaphragmal weakness, resp. failure[52]
PDK3	-	-	-	-	-	-	+ [74]	-	-	-
SURF1	-	-	-	-	-	-	-	-	-	-
DHTKD1	-	-	-	-	-	-	-	-	-	-
INF2	-	-	-	-	+ [65]	-	+ [65]	-	-	-
KIF1B	-	-	-	-	-	-	-	-	-	-
DYNC1HI	-	-	-	+ [77]	+ [77]	-	+ [77]	-	-	-
LMNA	-	-	-	+ [125]	-	-	-	-	-	-
NEFL	+[24]	+ [23]	+ [22]	-	-	+	+ [22]	-	-	-
TRIM2	-	-	-	-	-	-	+ [133]	-	-	-
GARS	-	-	-	-	-	-	-	-	-	-
KARS	-	-	-	-	-	-	+ [127]	-	-	-
AARS	-	-	-	-	-	-	-	-	-	-
MARS	-	-	-	-	+ [129]	-	-	-	-	-
HARS	-	-	-	-	+ [131]	-	-	-	-	-
YARS	-	-	-	-	-	-	-	-	-	-
HINT1	-	+ (ext)[132]	-	-	-	-	-	-	-	-
LRSAMI	-	+ (ext)[128]	-	-	-	-	-	-	-	-
SH3TC2	-	+ (to)[43]	+ [106]	+ [43]	+ [44]	-	+ [43]	pupill.[106]	-	resp. failure[107], left diaphragm paralysis[108]
RAB7	-	+ (ext)[111]	-	-	+ [50]	-	-	-	-	-
FIG4	-	-	+ [60]	+ [61]	+ [60]	+ [60]	+ [60]	ophthalmoparesis [60]	+ [115]	death due to resp. failure[116], elevated hemi-diaphragm[60]
DNM2	-	-	+ [107]	-	-	+ [108]	+ [119]	cataracts, ophthalmoparesis, strabismus[117]	-	-
MTMR2	-	-	+ [67]	+ [68]	-	-	+ [120]	-	-	death due to resp. failure[68], chronic stridor[67]
TFG	+[121]	+ (ext, tr, to)[69]	+ [69]	+ [69]	+ [69]	+ [122]	-	-	-	resp. failure[69]
LITAF	-	-	-	-	+ [71]	+ [123]	-	-	+	-
FGD4	-	-	-	-	-	-	+ [126]	pupill.[80]	-	-
SBF2	-	-	-	-	+ [86]	-	-	early-onset glaucoma[130]	-	-
NDRG1	-	-	+ [113]	-	-	-	+ [114]	pupill.[56]	-	-
HSPB8	-	-	-	-	-	-	-	-	-	-
HSPB1	-	+ (ext)[91]	-	-	+ [91]	-	-	-	-	-
HSII	-	-	-	-	-	+ [134]	-	-	-	-
MED25	-	-	-	-	-	+ [124]	-	-	-	-
IFRD1	+[92]	-	-	-	-	-	-	ophthalmoparesis[92]	-	-
TRPV4	-	-	+ [33]	+ [33]	-	-	+ [33]	strabismus[36], ophthalmoparesis [33], pupill.[36]	+ [36]	intercostal weakness, stridor, resp. failure[33]
SLC12A6	+[89]	-	+ [89]	-	-	-	+ [89]	strabismus[89]	-	-
HK1	-	-	+ [58]	+ [59]	-	-	-	-	-	-
AIFM1	-	+ [62]	-	-	-	-	+ [62]	-	-	-
PRPS1	-	-	-	-	-	-	-	optic atrophy bilat.[63]	-	-
FBLN5	-	-	-	-	-	+ [73]	-	macular degeneration[72]	+ [72]	resp. failure[72]
GNB4	-	-	-	-	-	-	-	-	-	-
SBFI	-	-	+ [85]	-	-	-	-	strabismus, pupill.[85]	-	-

Appendix S1 (continued): Additional symptoms and features in patients with CMT reported in the literature.

		[166]									
SH3TC2	+	-	-	VII-X + XII cranial nerve involvement[45], tongue atrophy + weakness[106]	unilateral brain atrophy [106]	+[43]	+[107]	-	-	-	-
RAB7	-	ulcers, infections, amputations, hypohidrosis [157], presyncopes[158]	+	nystagmus[50]	cerebellar degeneration [50]	-	-	-	-	-	-
FIG4	+[60]	-	-	tongue weakness[60]	cerebellar/ brain atrophy[60]	-	+[60]	-	-	-	involuntary movements, asymmetric weakness[60]
DNM2	-	-	-	-	-	-	-	-	-	+[117]	-
MTMR2	+[2]	-	+[68]	facial synkinesia[68], tongue atrophy, masticatory weakness[2]	-	-	+[163]	-	-	-	acrocyanosis [164]
TFG	-	constipation [69]	-	-	myelin pallor in spinal cord columns [121]	-	-	-	-	-	hyperglycemia, hyperlipidemia [69], myotonia, proximal>distal weakness, scapular + pelvic girdle atrophy[122]
LITAF	-	OH	-	-	-	-	+[165]	-	-	-	RLS
FGD4	-	-	+	[126]	-	-	+[80]	-	-	-	-
SBF2	-	-	-	cranial nerve involvement[87]	-	-	-	-	-	-	-
NDRG1	-	ulcers[113], bowel dysfunction[55]	-	nystagmus[160], tongue atrophy[113]	-	+[114]	+[161]	-	-	-	-
HSPB8	-	-	-	-	-	-	-	-	-	-	-
HSPB1	-	-	-	-	-	-	-	-	-	-	-
HSJ1	-	-	-	-	-	-	-	-	-	-	-
MED25	-	-	-	-	-	-	-	-	-	-	oedema[124]
IFRD1	-	-	-	coordination deficits, dysmetria, dysdiadochokinesia, nystagmus[92]	cerebellar atrophy[92]	-	-	-	-	-	proximal > distal weakness[92]
TRPV4	+[33]	U incontinence + urgency[39]	+[36]	abducens nerve palsy[156]	-	-	-	-	-	-	neck + trunk weakness[37], scapular winging[34]
SLC12A6	-	nocturnal vomiting[88]	+[89]	coordination deficits[88]	corpus callosum agenesis, enlarged ventricles, brain atrophy[89]	+[89]	-	-	-	-	epilepsy[89]
HK1	-	-	-	-	-	-	-	-	-	-	-
AIFM1	-	-	-	-	-	+[162]	-	-	-	-	-
PRPS1	-	-	-	-	-	-	-	-	-	-	-
FBLN5	-	chronic diarrhea[72]	+[72]	-	-	-	-	-	-	-	scapular winging, hyperelastic skin[72]
GNB4	-	-	-	-	-	-	-	-	-	-	-
SBF1	-	incontinence [85]	+[85]	-	brain atrophy[85]	-	-	-	-	-	-

Concerning PMP22: HNPP data are not included, cong. = congenital, CTS = carpal tunnel syndrome, pupill. = pupillary abnormality, resp. = respiratory, bilat. = bilateral, to = tongue, restr. = restrictive, ext = extremities, tr = trunk, SAS = sleep apnea syndrome, CNS = central nervous system, WM = white matter, MRI = magnetic resonance imaging, NCV = nerve conduction velocity, RLS = restless legs syndrome, ED = erectile dysfunction, OH = orthostatic hypotension, U = urinary, D = defecatory, FSGS = focal segmental glomerular sclerosis, ESRD = end stage renal disease, LL = lower limbs. Bold: new findings in current patient cohort; underlined: findings in current study that have been reported previously; not underlined and not bold: literature findings. Colors of the genes indicate their function within the peripheral nervous system. Yellow: myelination; light blue: mitochondrial; green: cytoskeletal stability and motor proteins; pink: RNA and protein metabolism; dark blue: protein folding; red: membrane traffic; grey: other/unknown. Violet: Transcription regulation. Brown: Channel/Transporter. Severe slow NCV < 10 m/s. Early onset < 18 month of age. Chest deformity = other than scoliosis.

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